

Collection of Family Health History for Assessment of Chronic Disease Risk in Primary Care

By: Karen P. Powell, Carol A. Christianson, Susan E. Hahn, Gaurav Dave, Leslie R. Evans, Susan H. Blanton, Elizabeth Hauser, Astrid Agbaje, Lori A. Orlando, Geoffrey S. Ginsburg, [Vincent C. Henrich](#)

Powell, K.P., Christianson, C.A., Hahn, S.E., Dave, G., Evans, L.R., Blanton, S.H., Hauser, E., Agbaje, A., Orlando, L.A., Ginsburg, G.S., Henrich, V.C. (2013). Collection of family health history for assessment of chronic disease risk in primary care. *North Carolina Medical Journal*, 74(4), 279-286.

Made available courtesy of the North Carolina Medical Society:

<http://www.ncmedicaljournal.com/>

*****© North Carolina Medical Society. Reprinted with permission. No further reproduction is authorized without written permission from the North Carolina Medical Society.*****

Keywords: Family Health History | Primary Care | Health Care Providers

*****Note: Full text of article below**

Collection of Family Health History for Assessment of Chronic Disease Risk in Primary Care

Karen P. Powell, Carol A. Christianson, Susan E. Hahn, Gaurav Dave, Leslie R. Evans, Susan H. Blanton, Elizabeth Hauser, Astrid Agbaje, Lori A. Orlando, Geoffrey S. Ginsburg, Vincent C. Henrich

BACKGROUND Family health history can predict a patient's risk for common complex diseases. This project assessed the completeness of family health history data in medical charts and evaluated the utility of these data for performing risk assessments in primary care.

METHODS Family health history data were collected and analyzed to determine the presence of quality indicators that are necessary for effective and accurate assessment of disease risk.

RESULTS More than 99% of the 390 paper charts analyzed contained information about family health history, which was usually scattered throughout the chart. Information on the health of the patient's parents was collected more often than information on the health of other relatives. Key information that was often *not* collected included age of disease onset, affected side of the family, and second-degree relatives affected. Less than 4% of patient charts included family health histories that were informative enough to accurately assess risk for common complex diseases.

LIMITATIONS Limitations of this study include the small number of charts reviewed per provider, the fact that the sample consisted of primary care providers in a single geographic location, and the inability to assess ethnicity, consanguinity, and other indicators of the informativeness of family health history.

CONCLUSIONS The family health histories collected in primary care are usually not complete enough to assess the patient's risk for common complex diseases. This situation could be improved with use of tools that analyze the family health history information collected and provide risk-stratified decision support recommendations for primary care.

Primary care providers routinely see patients who are at risk for, or are affected by, common complex diseases, such as coronary artery disease, cancer, and diabetes [1-5]. Family health history is one of the strongest predictors of the patient's risk for common complex diseases, and collecting this information can dramatically improve identification of at-risk individuals [4]. For example, having 1 first-degree relative with breast cancer results in a woman's risk for the disease being 1.8 times higher, and having 2 first-degree relatives nearly triples her risk [6]. Overall, 82% of primary care patients have a familial risk for at least 1 common disease—coronary artery disease, stroke, diabetes, breast cancer, colon cancer, or ovarian cancer [5, 7, 8]—and that risk alters the prevention recommendations for the patient. For instance, 15%–20% of patients meet family health history criteria for beginning colonoscopy screenings before age 50 years [9].

Because primary care providers are frequently a patient's first point of contact with the health care system, they are well positioned to identify patients who are at increased risk for disease and to implement appropriate prevention strategies in order to lower risk or detect disease earlier [2]. Professional [10-12] and evidence-based [13] guidelines are widely available; these guidelines can be used to collect family health histories for common diseases, such as colon cancer [14-17], breast cancer [17-20], heart disease [12], and

diabetes [21, 22]. Such guidelines have been endorsed by primary care organizations [10, 23, 24], yet they are underutilized in primary care settings [25-27]. Barriers to their use include the time required to collect an accurate family health history, the need to balance the patient's agenda with the physician's goals, the difficulty of finding information about family health history within the chart, and the lack of procedures for quickly collecting and analyzing family health history data [2, 28-31]. Using patient-collected information about family health history and incorporating it into the electronic medical record (EMR) might help to overcome these barriers [32, 33].

Making recommendations based on familial risk requires an accurate and detailed family health history [26]. Key elements of such a history are that it covers 3 generations (grandparents; parents, aunts, and uncles; and half siblings and full siblings) and that it includes age of disease onset, relationship to the patient, and age and cause of death (if deceased) for each individual [29, 34-36]. It is also impor-

Electronically published August 1, 2013.

Address correspondence to Ms. Karen Powell, University of North Carolina at Greensboro, 1111 Spring Garden St, Rm 3708, PO Box 26170, Greensboro, NC 27402 (Klpowell2@gmail.com).

N C Med J. 2013;74(4):279-286. ©2013 by the North Carolina Institute of Medicine and The Duke Endowment. All rights reserved. 0029-2559/2013/74402

tant that the family health history make note of common diseases that are *not* found in the family (a negative, or “unremarkable,” family history) [34]. Although most primary care providers collect a family health history, the documented elements vary [25, 26, 31].

The primary goal of this study was to assess the completeness of family health history data in the medical charts of primary care providers and to evaluate the utility of these data for providing patient risk assessments. The chart review was performed in selected primary care practices in a midsized community in the Southern United States.

Methods

Paper and EMR charts were reviewed in 1 internal medicine and 2 internal medicine/family medicine community-based practices serving patients with a range of socioeconomic and insurance statuses. The size of the practices ranged from 4 providers seeing 1,700 patients per month to 9 providers seeing 4,000 patients per month.

Data instrument. Our chart review checklist consists of 32 questions (including 14 multiple-choice questions, 9 dichotomous [yes/no] questions, and 9 fill-in-the-blank questions) and a chart on which the reviewer can circle whether particular types of information are “always,” “sometimes,” or “never” documented for various relatives. (See Appendix 1; online version only). The checklist is divided into sections dealing with the patient’s demographics and personal health history, the patient’s insurance, date(s) of personal and family health history collection, family health history data, and specialty referrals based on family health history. Fifteen of the questions on our checklist (marked with an asterisk) were adapted from the 2006 chart audit tool developed by the Michigan Department of Community Health’s genomics team (D. Duquette, unpublished observations, 2012). Our checklist was piloted and modified to ensure that all of the necessary information would be obtained.

APPENDIX 1. 2009–2010 Baseline Family History Chart Review

This appendix is available in its entirety in the online edition of this article. Please go to the NCMJ Web site <http://www.ncmedicaljournal.com/archives/?74402>.

The checklist and methodology were approved by the institutional review boards of the University of North Carolina at Greensboro, Moses H. Cone Memorial Hospital, and the US Army Medical Research and Materiel Command.

Sampling. Administrators from each practice provided a list of patients with outpatient paper charts, sorted by physician and appointment date, who were seen for a new visit or well visit between May 1 and November 1, 2007. In order to draw from patients throughout the list, every third chart

was reviewed, starting with the chart corresponding to a random digit provided by the Web site Random.org. At least 25 patient charts were reviewed for each provider. A chart was excluded if the patient was younger than 18 years or if the entire chart was not located onsite. Paper charts were abstracted by 2 genetic counselors between December 2008 and April 2010. Because 1 practice converted to an EMR system in 2010, an additional list of patients with outpatient EMR charts was provided; 2 study coordinators and the project director reviewed every other chart in this list in July and August of 2012. No identifying information was recorded, and standardized criteria for answering questions and interpreting family health histories were applied to reduce interobserver variability. Data quality was assessed by a genetics counselor who reviewed entries to correct errors. All data were analyzed in September 2012.

Statistical analysis. Abstracted data were entered into REDcap (Research Electronic Data Capture), a secure online survey and database storage tool. IBM SPSS Statistics software (version 19) was used for statistical analysis and reporting. Descriptive statistics were used to characterize the demographic characteristics of the patients and the health care providers and the characteristics of family health history collection.

To determine whether the family health history was informative enough to perform risk stratification and to alter a patient’s recommendations for prevention, this study used a set of quality indicators, which were subdivided by whether or not any of the patient’s family members were deceased. These quality indicators included: whether the family health history was updated during subsequent visits, whether a negative family health history was mentioned (eg, “no family health history of cancer”), whether the sex of affected relatives was noted, whether the age of the affected relative at disease onset was noted, and whether the affected relative’s lineage was noted (ie, whether the affected relative was on the maternal or paternal side of the family). Two additional quality indicators were used to assess family health history when at least 1 relative had died: the cause of death and the deceased relative’s age at death. Because each quality indicator is needed to perform an accurate risk assessment, family health histories were deemed highly informative (of high quality) only when the chart contained all 5 quality indicators (if the chart made no mention of any relative being deceased) or all 7 quality indicators (if any relative was deceased).

Results

A total of 399 paper charts and 100 EMR charts were abstracted and entered into REDcap. Data from the paper charts were cleaned and corrected when necessary, resulting in the removal of 9 records.

Patient and Physician Characteristics

Physician characteristics are presented in Table 1, and patient characteristics are presented in Table 2. The median

TABLE 1.
Characteristics of Providers Whose Patient Charts Were Reviewed (N = 16)

Characteristic	Number of providers (%)
Sex	
Male	4 (25.0)
Female	12 (75.0)
Years in practice	
≤21 years	8 (50.0)
>21 years	8 (50.0)
Medical specialty	
Family medicine	7 (43.8)
Internal medicine	9 (56.3)
Race	
White	13 (81.3)
Asian	2 (12.5)
Hispanic	1 (6.3)

patient age was 53 years. The most commonly noted diseases were cardiovascular disease (CVD) and cancer. In personal disease histories, hypercholesterolemia was the most frequently mentioned type of CVD; it was noted in the charts of 180 (76.3%) of the 236 patients with CVD. Nonmelanoma skin cancer was the most commonly noted form of cancer; it was reported in 27 (55.1%) of the 49 patients with cancer. The average number of years a patient had been seen in the practice was 9.76 (± 8.38) years, and the median length of time a patient had been seen in the practice was 8 years.

Family Health History Within Paper Charts

Location. More than 99% of paper charts contained some family health history data, which was scattered over several areas of the chart. In 306 of the 390 charts (78.5%), family health history was located in the physician's notes. In 280 (71.8%) of the charts, the family health history was found on the patient's self-completed intake form; in 97 charts (24.9%), it was on the front summary page of the chart; in 63 charts (16.2%) it was found in the consult notes; in 16 charts (4.1%) it was found in a note from the patient; and in 1 (0.3%) of the charts, it was found in a nurse's note. Frequently, family health history was noted separately in 2 places. None of the examined charts contained a family health history in pedigree format.

Relatives assessed. When we looked at the health history of affected family members, we found that the health history of the patient's parents was documented in 339 (86.9%) of 390 charts, while only about half as many charts (168 [43.1%]) contained the health history of siblings. Only 131 charts (33.6%) contained the health history of grandparents; 67 charts (17.2%) contained the health history of aunts or uncles; and 32 charts (8.2%) contained the health history of children.

Diseases collected. A total of 390 charts were reviewed to assess whether the patient's family health history mentioned either the presence of a disease (positive history) or

the absence of a disease (negative history); we looked for diseases such as CVD, cancer, stroke, diabetes mellitus, arthritis, or depression. For example, more than three-quarters of charts had a positive family history of CVD, whereas only 23.3% of the charts mentioned the absence of CVD in the family health history. Table 3 shows the diseases and conditions for which data were collected. Table 4 shows the number and proportion of charts that recorded each of the quality indicators mentioned previously; these quality indicators are also discussed below.

Quality indicators. One quality indicator is whether the family health history has been updated. After an initial visit, the number of years before the first family health history was recorded in a patient's chart ranged from 0 to 32 years, with a median of 0.0 and an interquartile range of 1.00. On average, the most recent family health history had been collected or updated within the past 0.18 years (standard deviation = 1.06). Of the 390 charts reviewed, 287 charts (73.6%) had been updated; 180 (62.7%) of these charts had all updated changes, 53 (18.5%) of them had some updated changes, and 54 charts (18.8%) indicated that patients had been asked about updates but no changes had been made.

TABLE 2.
Characteristics of Patients Whose Charts Were Reviewed (N = 390)

Characteristic	Number of patients (%)
Sex	
Female	200 (51.3)
Male	187 (47.9)
Missing data	3 (0.8)
Race	
White	251 (64.3)
African American	59 (15.1)
Hispanic	4 (1.0)
Asian	4 (1.0)
Other	4 (1.0)
Missing data	68 (17.6)
Type of insurance	
Commercial	269 (68.9)
Medicare	54 (13.8)
Medicaid	4 (1.0)
Self-pay	1 (0.2)
Unable to determine	55 (14.1)
Missing data	7 (1.7)
Medical conditions	
Cardiovascular diseases	236 (60.5)
Hypercholesterolemia	180 (46.1)
Hypertension	157 (40.2)
Other cardiovascular diseases	28 (7.1)
Cancer	49 (12.5)
Skin cancer	27 (6.9)
Breast cancer	11 (2.8)
Prostate cancer	6 (1.5)
Other type of cancer	5 (1.2)

TABLE 3.
Types of Family Health History Identified in Reviewed Charts^a (N = 390)

Medical conditions of relatives	Charts with positive FHH Number of charts (%)	Charts with negative FHH Number of charts (%)
Cardiovascular diseases	338 (86.7)	91 (23.3)
Hypertension	242 (62.0)	22 (5.6)
Heart attack	150 (38.4)	10 (2.5)
Hypercholesterolemia	69 (17.6)	4 (1.1)
Cancer	266 (68.2)	195 (50.0)
Breast cancer	83 (21.2)	34 (8.8)
Colon cancer	67 (17.2)	62 (15.9)
Lung cancer	64 (16.4)	0 (0)
Stroke	71 (18.2)	4 (1.1)
Other conditions		
Diabetes	200 (51.2)	48 (12.4)
Arthritis	71 (18.2)	14 (3.6)
Depression	39 (10.0)	12 (3.1)

Note. FHH, family health history.

^aTotals do not sum to the sample size because of missing data.

Of the histories that were being taken for the first time, 37 (66.1%) were for patients who were new to the practice.

Another quality indicator is whether a negative family health history is reported. Almost half (173 [44.4%]) of the charts explicitly recorded a generalized negative statement regarding family health history for a specific disease or disease group (eg, "family history negative for cancer").

A third quality indicator is whether the sex of the affected relative is reported in the family health history. In 366 (93.8%) of the charts, a positive family history of a specific disease or disease group was noted. In these charts, the sex of the affected relative was the most frequently collected quality indicator, having been specified in 356 (91.2%) of the charts reviewed. In some cases, the sex of the affected relative was known because of the words used to describe the relative (ie, aunt, uncle, mother, father, sister, brother). The sex of the affected relative was noted in 92% of the instances in which the relative was a parent, aunt, uncle, sibling, or grandparent. The sex of the affected relative was noted in only 3 (17.6%) of the instances in which the relative was a cousin and in only 6 (24.0%) of the instances in which the individual was described as a "relative."

Age at disease onset for an affected relative was the least frequently collected quality indicator, having been collected in only 71 (18.2%) of the 366 family health histories that recorded a positive family history. Specifically, age of disease onset was documented in family health histories for 11 (6.6%) of the siblings mentioned, 14 (10.8%) of the grandparents mentioned, 4 (6.3%) of the aunts or uncles mentioned, and 50 (14.8%) of the parents mentioned.

The fifth quality indicator for a family health history is whether the lineage of the affected relative is reported. Of the 366 family health histories that recorded a positive family history, 255 (69.7%) did not include information about

the lineage (ie, maternal or paternal side) of affected family members (Table 4). More than half of the 366 charts (233 [63.7%]) did not mention an affected second-degree relative, and 44 charts did not mention an affected first-degree relative.

If the family health history includes mention of deceased relatives, then 2 additional quality indicators should be evaluated: age at death and cause of death. A deceased relative was documented in 227 (62.0%) of the 366 records with a positive family health history. In 172 (75.8%) of those 227 records, the affected relative's age at death was recorded, either for all deceased relatives (94/227 [41.4%]) or for some of them (78/227 [34.4%]). Of the 227 charts that noted a deceased relative, 213 (93.8%) listed the cause of death, either for all deceased relatives (165/227 [72.7%]) or for some of them (48/227 [21.1%]).

Quality of family health history. Less than 4% of patients had family health histories that could be used to perform a risk assessment. The group of 227 family health histories that mentioned a deceased relative included more "moderately informative" histories and fewer "less informative" histories than did the group of family histories that did not include mention of any deceased relatives (Table 4). The number of quality indicators present in each group is shown in Table 4, and Table 5 shows the number of charts in which each of the first 5 quality indicators was reported. Among the charts that did not mention any deceased relative, 61 charts included 4 of the 5 quality indicators; the indicators that were most frequently absent were the age of the affected relative (missing in 28 [45.9%] of the charts), negative family health history information (missing in 16 [26.2%] of the charts), and the lineage of the affected relative (missing in 15 [24.6%] of the charts). Similar results were observed for the charts that mentioned one or more deceased relatives.

TABLE 4.
Characteristics of Reviewed Charts in Family Health History Project

Characteristic	All charts (N = 390) Number of charts (%)	Charts with FHH that do not mention deceased relatives (n = 163) Number of charts (%)	Charts with FHH that do mention deceased relatives (n = 277) Number of charts (%)
FHH status			
Updated	287 (73.5)	92 (56.4)	186 (81.9)
Not updated	91 (23.3)	49 (30.0)	38 (16.7)
Missing data	12 (3.2)	22 (13.6)	3 (1.4)
Negative FHH			
Recorded	173 (44.3)	70 (42.9)	92 (40.5)
Not recorded	213 (54.6)	71 (43.5)	135 (59.5)
Missing data	4 (1.1)	22 (13.6)	0 (0)
Affected relative^a			
Recorded	366 (93.8)	135 (82.8)	226 (99.6)
Not recorded	24 (6.2)	6 (3.6)	1 (0.4)
Missing data	0 (0)	22 (13.6)	0 (0)
Sex of affected relative			
Recorded	356 (91.2)	129 (79.1)	222 (97.8)
Not recorded	10 (2.5)	12 (7.3)	5 (2.2)
Missing data	24 (6.3)	22 (13.6)	0 (0)
Affected relative's age at disease onset			
Recorded	71 (18.2)	27 (16.5)	44 (19.3)
Not recorded	256 (65.6)	108 (66.2)	182 (80.1)
Missing data	63 (16.2)	28 (17.3)	1 (0.6)
Lineage of affected relative			
Recorded	111 (28.4)	50 (30.6)	58 (25.6)
Not recorded	255 (65.3)	91 (55.2)	169 (74.4)
Missing data	24 (6.3)	22 (14.2)	0 (0)
Cause of death of affected relative			
Recorded	NA	NA	213 (93.8)
Not recorded	NA	NA	14 (6.2)
Age of affected relative at death			
Recorded	NA	NA	172 (75.8)
Not recorded	NA	NA	55 (24.2)
Number of quality indicators present			
0	2 (0.6)	2 (1.5)	0 (0)
1	37 (10.2)	14 (10.4)	2 (0.9)
2	119 (32.9)	41 (30.4)	6 (2.7)
3	131 (36.2)	49 (36.3)	32 (14.3)
4	61 (16.9)	26 (19.3)	86 (38.6)
5	12 (3.3)	3 (2.2)	65 (29.1)
6	NA	NA	24 (10.8)
7	NA	NA	8 (3.6)
Quality of FHH			
Not informative ^b	2 (0.6)	2 (1.5)	0 (0)
Less informative ^c	156 (43.0)	55 (40.7)	40 (18.0)
Moderately informative ^d	192 (53.1)	75 (55.6)	175 (78.4)
Highly informative ^e	12 (3.3)	3 (2.2)	8 (3.6)

Note. FHH, family health history; NA, not applicable.

^a"Not recorded" means a characteristic was not asked about or was not checked in the chart; "missing data" means data is completely missing from the dataset.

^bNot included as an indicator of quality.

^cPedigrees were labeled "not informative" if they contained none of the quality indicators.

^dPedigrees were labeled "less informative" if they did not mention any deceased relatives and contained 1-2 quality indicators, or if they did mention a deceased relative and contained 1-3 quality indicators.

^ePedigrees were labeled "moderately informative" if they did not mention any deceased relatives and contained 3-4 quality indicators, or if they did mention a deceased relative and contained 4-6 quality indicators.

^fPedigrees were labeled "highly informative" if they did not mention any deceased relatives and contained 5 quality indicators, or if they did mention a deceased relative and contained 7 quality indicators.

TABLE 5.
Number of Charts Containing Each Type of Quality Indicator, Cross-Tabulated with the Number of Quality Indicators Present in the Chart (Number of Charts = 390)

Quality indicator	Number of indicators present				
	1	2	3	4	5
FHH status updated	3	91	112	59	12
Negative family history recorded	2	20	82	45	12
Sex of affected relative recorded	32	116	131	61	12
Affected relative's age at disease onset recorded	0	3	23	33	12
Lineage of affected relative recorded	0	8	45	46	12

Note. FHH, family health history.
 Totals do not sum to the sample size because of missing data.

Family Health History Within EMRs

Out of 100 EMR charts, 97 (97%) documented some amount of family health history. No patient had a structured 3-generation pedigree. Interestingly, the EMR's family health history collection tool was not utilized for any of the charts we reviewed. In all cases, family health history was recorded in the free text section of the clinic note or on the patient intake form, and family health history was included for only a select few relatives.

Discussion

The inability to use family health histories in primary care poses a barrier to the practice of genomic medicine and limits physicians' ability to achieve benchmarks set by programs such as Healthy People 2020 [37]. Several problems were encountered with the charts analyzed in this study, including a lack of uniformity in the collection methods, variations in the location within the chart where family health history information was reported, and missing information about essential elements of the family health history (as presented in Tables 4 and 5). Without all of these elements, an adequate risk assessment cannot be performed.

Family health histories that included information about deceased relatives were more likely to be "moderately informative" than were those without any mention of deceased relatives. The algorithm used to assess the quality of the family health histories for the deceased-relative group included 2 additional indicators (age at death and cause of death of the affected relative). More than two-thirds of the family health histories in the deceased-relative group recorded these 2 indicators, thereby raising the mean quality of the family health histories and the quality of the information. It may be that when the death of an affected relative is recorded in a family health history, health care providers are more likely to seek details on the age at death and cause of death.

Although more than 97% of family health histories were updated, almost 23% contained incomplete information. Missing information included information that was previ-

ously collected but was not brought forward during subsequent visits and was thus lost. Some new EMR programs have addressed this problem.

In 80%-90% of the charts we reviewed, the age at diagnosis of an affected relative was never mentioned. Diagnosis of, or death from, a disease at an earlier age than expected can indicate a hereditary form of disease and is included in clinical algorithms to evaluate a patient's disease risk [13, 14, 19, 38]. Thus, there is a need for greater awareness about the importance of age at diagnosis for risk assessment. Age at diagnosis is particularly important when using family health history to identify those most at risk for heart disease and cancer, the 2 leading causes of death in the United States [39]. Interestingly, using an EMR did not increase the amount or quality of information collected by the practitioner. Because EMRs presently do not prompt the physician to collect all of the elements necessary for an assessment of disease risk based on family health history, it remains uncertain whether family health histories will be fully utilized for these widespread diseases.

The family health histories of cancer documented in these charts resemble those found in other studies [5, 31]. The vast majority of patient charts had a documented positive family health history; in more than 45% of cases, however, diseases that were not found in the family (ie, negative family history) were not explicitly mentioned. If a negative family history was mentioned by a consulting physician, it typically was not updated in the primary care provider's chart and was not considered when assessing the patient's risk for disease. Documentation of a negative family health history is almost as important as documentation of a positive family health history, and primary care providers may benefit from greater awareness of its value for interpreting family health history data [34].

Several studies have estimated the chance of having an increased risk for disease on the basis of the family health history taken by the patient's primary care provider [7, 8]. Based on the estimated frequency of at-risk patients seen in previous studies, we would have expected approximately 180 individuals in this study to have a family health history that established a strong or moderate risk for breast and ovarian cancer or colon cancer, which would suggest the need for referral to a genetic specialist. However, no such referrals were made. Although patients were sometimes referred to a specialist, such as an oncologist or a gastroenterologist, charts rarely mentioned that a patient was at high risk for disease based on family health history; this is probably because such an evaluation would be time-consuming and difficult for the primary care provider, even if the family health history were accurate and complete. Most charts had a patient intake form that allowed patients to self-report family health history. Several studies have found a bias for patients to overreport or underreport diseases, specifically cancer [40-42]. This may be another challenge to the effective utilization of family health history information.

Overcoming barriers to collection and use of family health histories in primary care. To remove several of the deficiencies we observed in the collection and interpretation of family health histories, it would be helpful if clinicians had a tool that creates a structure containing all of the key elements of family health histories (eg, a pedigree), provides decision support for providers [5, 32, 43], and is compatible with EMRs. A family health history decision support tool would need to be able to do the following things: allow patients to fill out a form about their family health history using a secure online system prior to their appointment; update family health histories without dropping previously collected information about family members; provide a single location for family health history within a chart (especially if the tool is compatible with the EMR); easily identify the number of affected and deceased family members; allow providers to quickly see whether a family has a negative disease history; and use clinically established information to provide an assessment of disease risk and recommendations regarding medical management.

Limitations

Limitations of this study that may reduce the generalizability of its findings to other populations include the small number of charts reviewed per primary care provider, the fact that the sample of primary care providers were all from a single geographic location, the failure to differentiate between patient-generated and physician-collected family health histories, and the inability to assess other quality indicators, such as consanguinity. The latter limitation results from confusion about the information included in charts. For example, a chart might be unclear as to whether the number of family members in the family matches the number of relatives mentioned in the family health history (eg, a patient might have 3 siblings, only 1 of whom is mentioned in the family health history), or there might be uncertainty about some pieces of information (eg, the family health history might mention a brother with heart disease and later refer to a brother with diabetes, without explaining whether these are 2 references to the same person or references to 2 different brothers). Another problem is that the chart review checklist does not collect certain additional types of information (eg, consanguinity). Lastly, there is evidence in the literature that patient-provided family health histories can differ from physician-collected histories. Most charts contained a patient intake form that had been used to collect family health history data, and this form was considered part of the family health history in the patient record.

Despite these limitations, our findings are consistent with those of previous studies, suggesting common factors in the use of family health histories across multiple settings. This study also did not allow for assessment of conversations between patients and providers; therefore, any information about family health history that was discussed in these conversations but was not mentioned in the clinic

notes would not have been collected. The impact of verbally communicated but undocumented family health history information is unknown but is important to consider, especially in primary care settings where relationships are often long-lasting and the time available for documentation is often limited.

Conclusion

Despite the value of family health histories for disease risk assessment, the histories collected in primary care practices usually lack some of the crucial information needed to perform a risk assessment for hereditary cancer syndromes and other chronic disorders [26]. If the use of family health histories for disease risk assessment is to succeed in primary care, collection must be easier and more complete. This study indicates a need for the adoption of family health history collection tools that can analyze the collected information and provide risk-stratified decision support recommendations. *NCMJ*

Karen P. Powell, MS project coordinator/genetic counselor, Cone Cancer Center, Greensboro, North Carolina.

Carol A. Christianson, MS genetic counselor, Cancer Genetics Program, West Michigan Cancer Center, Kalamazoo, Michigan.

Susan E. Hahn, MS assistant director of communications, compliance and ethics, John P. Hussman Institute for Human Genomics, University of Miami, Miami, Florida.

Gaurav Dave, MD, DrPH, MPH assistant director, NC TraCS Institute, University of North Carolina at Chapel Hill, Chapel Hill, North Carolina.

Leslie R. Evans, MS genetic counselor, Center for Biotechnology, Genomics and Health Research, University of North Carolina at Greensboro, Greensboro, North Carolina.

Susan H. Blanton, PhD associate director of communications and compliance, and executive director, John P. Hussman Institute for Human Genomics, and associate professor, Dr. John T. Macdonald Foundation Department of Human Genetics, University of Miami, Miami, Florida.

Elizabeth Hauser, PhD professor, Department of Medicine and Center for Human Genetics, Duke University, Durham, North Carolina.

Astrid Agbaje, MA project director, Cone Health, Greensboro, North Carolina.

Lori A. Orlando, MD, MHS assistant professor of medicine, Department of Medicine and Center for Personalized Medicine, Duke University, Durham, North Carolina.

Geoffrey S. Ginsburg, MD, PhD director, Institute for Genome Sciences and Policy; executive director, Center for Personalized and Precision Medicine; and professor of medicine and pathology, Duke University School of Medicine, Durham, North Carolina.

Vincent C. Henrich, PhD director, Center for Biotechnology, Genomics, and Health Research; and professor of biology, University of North Carolina at Greensboro, Greensboro, North Carolina.

Acknowledgments

The Genomical Connection is an alliance between the University of North Carolina at Greensboro, Duke University, and Cone Health to promote improved uptake and use of family health history and genomics for risk assessment and disease prevention.

Financial support. Funding for this project was provided by the US Department of the Army (W81XWH-05-1-0383).

Potential conflicts of interest. K.P. consults with CareCore National and Generation Health. C.C. has consulted with CareCore National. L.E. has been employed by, and owns stock in, Counsyl and is currently employed by Life Technologies. G.G. consults for CardioDx, Universal Oncology, and BG Medicine; owns stock in CardioDx and Universal Oncology; has 2 current grants each with Novartis and Pfizer; has received 3 patents and has 2 pending patents; and receives royalties from Elsevier. V.H. consults for LabCorp. All other authors have no conflicts of interest.

References

- Feero WG. Genetics of common disease: a primary care priority aligned with a teachable moment? *Genet Med*. 2008;10(2):81-82.
- Guttmacher AE, Jenkins J, Uhlmann WR. Genomic medicine: who will practice it? A call to open arms. *Am J Med Genet*. 2001;106(3):216-222.
- Burke W, Emery J. Genetics education for primary-care providers. *Nat Rev Genet*. 2002;3(7):561-566.
- Qureshi N, Armstrong S, Dhiman P, et al. Effect of adding systematic family history enquiry to cardiovascular disease risk assessment in primary care: a matched-pair, cluster randomized trial. *Ann Intern Med*. 2012;156(4):253-262.
- O'Neill SM, Rubinstein WS, Wang C, et al. Familial risk for common diseases in primary care: the Family Healthcare Impact Trial. *Am J Prev Med*. 2009;36(6):506-514.
- American Cancer Society. Breast Cancer Facts and Figures 2011-2012. Atlanta, GA: American Cancer Society Inc; 2011. <http://www.cancer.org/acs/groups/content/@epidemiologysurveillance/documents/document/acspc-030975.pdf>. Accessed June 2, 2013.
- Scheuner MT, Whitworth WC, McGruder H, Yoon PW, Khoury MJ. Familial risk assessment for early-onset coronary heart disease. *Genet Med*. 2006;8(8):525-531.
- Ramsey SD, Yoon P, Moonesinghe R, Khoury MJ. Population-based study of the prevalence of family history of cancer: implications for cancer screening and prevention. *Genet Med*. 2006;8(9):571-575.
- Winawer SJ, Fletcher RH, Miller L, et al. Colorectal cancer screening: clinical guidelines and rationale. *Gastroenterology*. 1997;112(2):594-642.
- American College of Obstetrics and Gynecology Committee on Genetics. Committee Opinion No. 478: Family history as a risk assessment tool. *Obstet Gynecol*. 2011;117(3):747-750.
- Pletcher BA, Toriello HV, Noblin SJ, et al. Indications for genetic referral: a guide for healthcare providers. *Genet Med*. 2007;9(6):385-389.
- Greenland P, Alpert JS, Beller GA, et al. 2010 ACCF/AHA guideline for assessment of cardiovascular risk in asymptomatic adults: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines. *Circulation*. 2010;122(25):e584-e636.
- Scheuner MT, Wang SJ, Raffel LJ, Larabell SK, Rotter JI. Family history: a comprehensive genetic risk assessment method for the chronic conditions of adulthood. *Am J Med Genet*. 1997;71(3):315-324.
- National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology. NCCN Guidelines for Detection, Prevention, and Risk Reduction. Colorectal Cancer Screening. NCCN Web site. http://www.nccn.org/professionals/physician_gls/f_guidelines.asp#detection. Accessed June 2, 2013.
- US Preventive Services Task Force. Screening for colorectal cancer: U.S. Preventive Services Task Force Recommendation Statement. *Ann Intern Med*. 2008;149(9):627-637.
- Robson ME, Storm CD, Weitzel J, Wollins DS, Offit K; American Society of Clinical Oncology. American Society of Clinical Oncology policy statement update: genetic and genomic testing for cancer susceptibility. *J Clin Oncol*. 2010;28(5):893-901.
- Smith R, Cokkinides V, Brauley OW. Cancer screening in the United States, 2008: a review of current American Cancer Society guidelines and cancer screening issues. *CA Cancer J Clin*. 2008;58(3):161-179.
- Daly MB, Axilbund JE, Buys S, et al. Genetic/familial high-risk assessment: breast and ovarian—clinical practice guidelines in oncology. *J Natl Compr Canc Netw*. 2010;8(5):562-594.
- National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology. NCCN Guidelines for Detection, Prevention, and Risk Reduction. Genetic/Familial High-Risk Assessment: Breast and Ovarian. NCCN Web site. http://www.nccn.org/professionals/physician_gls/f_guidelines.asp#detection. Accessed June 2, 2013.
- Humphrey LL, Helfand M, Chan BK, Woolf SH. Breast cancer screening: a summary of the evidence for the U.S. Preventive Services Task Force. *Ann Intern Med*. 2002;137(5 pt 1):347-360.
- Hariri S, Yoon PW, Qureshi N, Valdez R, Scheuner MT, Khoury MJ. Family history of type 2 diabetes: a population-based screening tool for prevention? *Genet Med*. 2006;8(2):102-108.
- Valdez R, Yoon PW, Liu T, Khoury MJ. Family history and prevalence of diabetes in the U.S. population: the 6-year results from the National Health and Nutrition Examination Survey (1999-2004). *Diabetes Care*. 2007;30(10):2517-2522.
- American Academy of Family Physicians Core Educational Guidelines. Medical genetics: recommended core educational guidelines for family practice residents. *Am Fam Physician*. 1999;60(1):305-307.
- American Academy of Family Physicians. Recommended Curriculum Guidelines for Family Medicine Residents: Medical Genetics. AAFP Web site. http://www.aafp.org/online/etc/medialib/aafp_org/documents/about/rap/curriculum/medical_genetics.Par0001.File.tmp/medical-genetics.pdf. Revised June 2012 by Atlantic Health System. Accessed June 2, 2013.
- Acheson LS, Wiesner GL, Zyzanski SJ, Goodwin MA, Stange KC. Family history-taking in community family practice: implications for genetic screening. *Genet Med*. 2000;2(3):180-185.
- Murff HJ, Byrne D, Syngal S. Cancer risk assessment: quality and impact of the family history interview. *Am J Prev Med*. 2004;27(3):239-245.
- Frezzo TM, Rubinstein WS, Dunham D, Ormond KE. The genetic family history as a risk assessment tool in internal medicine. *Genet Med*. 2003;5(2):84-91.
- Suther S, Goodson P. Barriers to the provision of genetic services by primary care physicians: a systematic review of the literature. *Genet Med*. 2003;5(2):70-76.
- Guttmacher AE, Porteous ME, McInerney JD. Educating health-care professionals about genetics and genomics. *Nat Rev Genet*. 2007;8(2):151-157.
- Sabatino SA, McCarthy EP, Phillips RS, Burns RB. Breast cancer risk assessment and management in primary care: provider attitudes, practices, and barriers. *Cancer Detect Prev*. 2007;31(5):375-383.
- Christianson CA, Powell KP, Hahn SE, Blanton SH, Bogacik J, Henrich VC; Genomical Connection. The use of a family history risk assessment tool in a community health care system: views of primary care providers. *J Genet Couns*. 2012;21(5):652-661.
- Orlando LA, Hauser ER, Christianson C, et al. Protocol for implementation of family health history collection and decision support into primary care using a computerized family health history system. *BMC Health Serv Res*. 2011;11:264.
- Giovanni MA, Murray MF. The application of computer-based tools in obtaining the genetic family history. *Curr Protoc Hum Genet*. 2010;Chapter 9:Unit9.21.
- Bennett RL. The family medical history. *Prim Care*. 2004;31(3):479-495, vii-viii.
- Qureshi N, Carroll JC, Wilson B, et al. The current state of cancer family history collection tools in primary care: a systematic review. *Genet Med*. 2009;11(7):495-506.
- Berg AO, Baird MA, Botkin JR, et al. National Institutes of Health State-Of-The-Science Conference Statement: Family History and Improving Health. *Ann Intern Med*. 2009;151(12):872-877.
- Healthy People 2020. Healthy People 2020: Topics and Objectives. HealthyPeople.gov Web site. <http://www.healthypeople.gov/2020/topicsobjectives2020/>. Last updated March 8, 2013. Accessed June 5, 2013.
- Hunt SC, Williams RR, Barlow GK. A comparison of positive family history definitions for defining risk of future disease. *J Chronic Dis*. 1986;39(10):809-821.
- Centers for Disease Control and Prevention (CDC) and National Center for Health Statistics. FastStats. Leading Causes of Death. Final 2010 data. CDC Web site. <http://www.cdc.gov/nchs/fastats/lcod.htm>. Last updated January 11, 2013. Accessed June 4, 2013.
- Ozanne EM, O'Connell A, Bouzan C, et al. Bias in the reporting of family history: implications for clinical care. *J Genet Couns*. 2012;21(4):547-556.
- Orom H, Coté ML, González HM, Underwood W 3rd, Schwartz AG. Family history of cancer: is it an accurate indicator of cancer risk in the immigrant population? *Cancer*. 2008;112(2):399-406.
- Quillin JM, Ramakrishnan V, Borzelleca J, Bodurtha J, Bowen D, Baer Wilson D. Paternal relatives and family history of breast cancer. *Am J Prev Med*. 2006;31(3):265-268.
- Yoon PW, Scheuner MT, Jorgensen C, Khoury MJ. Developing Family Healthcare, a family history screening tool to prevent common chronic diseases. *Prev Chronic Dis*. 2009;6(1):A33.